



Wolman disease

Wolman disease is a rare inherited condition involving the breakdown and use of fats and cholesterol in the body (lipid metabolism). In affected individuals, harmful amounts of lipids accumulate in the spleen, liver, bone marrow, small intestine, small hormone-producing glands on top of each kidney (adrenal glands), and lymph nodes. In addition to fat deposits, calcium deposits in the adrenal glands are also seen.

Infants with Wolman disease are healthy and active at birth but soon develop signs and symptoms of the disorder. These may include an enlarged liver and spleen (hepatosplenomegaly), poor weight gain, low muscle tone, a yellow tint to the skin and the whites of the eyes (jaundice), vomiting, diarrhea, developmental delay, low amounts of iron in the blood (anemia), and poor absorption of nutrients from food. Children affected by this condition develop severe malnutrition and generally do not survive past early childhood.

Frequency

Wolman disease is estimated to occur in 1 in 350,000 newborns.

Genetic Changes

Mutations in the *LIPA* gene cause Wolman disease.

The *LIPA* gene provides instructions for producing an enzyme called lysosomal acid lipase. This enzyme is found in the lysosomes (compartments that digest and recycle materials in the cell), where it processes lipids such as cholesteryl esters and triglycerides so they can be used by the body.

Mutations in this gene lead to a shortage of lysosomal acid lipase and the accumulation of triglycerides, cholesteryl esters, and other kinds of fats within the cells and tissues of affected individuals. This accumulation as well as malnutrition caused by the body's inability to use lipids properly result in the signs and symptoms of Wolman disease.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- Acid lipase deficiency
- Familial Xanthomatosis
- LAL deficiency
- LIPA deficiency
- Liposomal Acid Lipase Deficiency, Wolman Type
- Lysosomal acid lipase deficiency

Diagnosis & Management

These resources address the diagnosis or management of Wolman disease:

- Genetic Testing Registry: Lysosomal acid lipase deficiency
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0043208/>
- National Organization for Rare Disorders (NORD) Physician Guide
<http://nordphysicianguides.org/lipoprotein-lipase-deficiency-lpld/>

These resources from MedlinePlus offer information about the diagnosis and management of various health conditions:

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>

Additional Information & Resources

MedlinePlus

- Health Topic: Lipid Metabolism Disorders
<https://medlineplus.gov/lipidmetabolismdisorders.html>

Genetic and Rare Diseases Information Center

- Lysosomal acid lipase deficiency
<https://rarediseases.info.nih.gov/diseases/12097/lysosomal-acid-lipase-deficiency>
- Wolman disease
<https://rarediseases.info.nih.gov/diseases/7899/wolman-disease>

Additional NIH Resources

- National Institute of Neurological Disorders and Stroke: Acid Lipase Disease Information Page
<https://www.ninds.nih.gov/Disorders/All-Disorders/Acid-Lipase-Disease-Information-Page>
- National Institute of Neurological Disorders and Stroke: Lipid Storage Diseases Fact Sheet
<https://www.ninds.nih.gov/Disorders/All-Disorders/Lipid-storage-diseases-Information-Page>

Educational Resources

- Disease InfoSearch: Wolman disease
<http://www.diseaseinfosearch.org/Wolman+disease/7523>
- MalaCards: wolman disease
http://www.malacards.org/card/wolman_disease
- My46 Trait Profile
<https://www.my46.org/trait-document?trait=Lysosomal%20acid%20lipase%20deficiency&type=profile>
- Orphanet: Wolman disease
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=75233

Patient Support and Advocacy Resources

- CLIMB: Children Living with Inherited Metabolic Diseases
<http://www.climb.org.uk/>

Genetic Testing Registry

- Lysosomal acid lipase deficiency
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0043208/>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22Wolman+disease%22+OR+%22Wolman+Disease%22>

Scientific articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28Wolman+Disease%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- LYSOSOMAL ACID LIPASE DEFICIENCY
<http://omim.org/entry/278000>

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